



DVL1 gene

dishevelled segment polarity protein 1

Normal Function

The *DVL1* gene provides instructions for making a protein that plays a critical role in development starting before birth. The DVL1 protein participates in chemical signaling pathways known as WNT signaling. These pathways control the activity of certain genes and regulate the interactions between cells during embryonic development. Signaling involving the DVL1 protein appears to be important for the normal development of the brain, skeleton, and many other parts of the body.

Health Conditions Related to Genetic Changes

Robinow syndrome

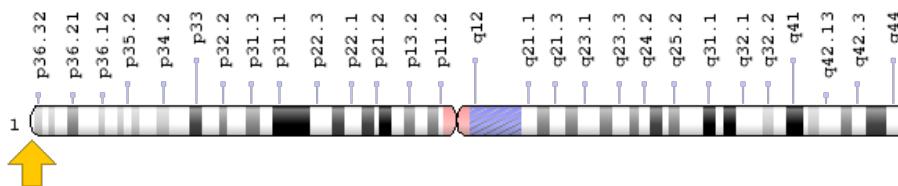
At least six mutations in the *DVL1* gene have been found to cause the autosomal dominant form of Robinow syndrome, a condition that affects the development of many parts of the body, particularly the bones. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. *DVL1* gene mutations underlie a variant type of this condition called the osteosclerotic form. Signs and symptoms of the osteosclerotic form include increased bone mineral density (osteosclerosis) in addition to the usual features of Robinow syndrome.

All of the identified *DVL1* gene mutations occur in a region of the gene known as exon 14. Each mutation is predicted to remove a segment of protein building blocks (amino acids) from one end of the DVL1 protein and add more than 100 new amino acids. Researchers are working to determine how these structural changes affect the protein's function. The changes may have a dominant-negative effect, which would mean that the altered protein produced from one copy of the *DVL1* gene interferes with the function of the normal protein produced from the other copy of the gene. Alternately, the changes may have a gain-of-function effect, giving the altered protein a new, as-yet-undetermined function. Either way, the altered protein likely impairs WNT signaling. Problems with these signaling pathways disrupt the normal development of many organs and tissues, leading to the features of Robinow syndrome. It is unclear why *DVL1* gene mutations cause osteosclerosis in addition to the usual signs and symptoms of the condition.

Chromosomal Location

Cytogenetic Location: 1p36.33, which is the short (p) arm of chromosome 1 at position 36.33

Molecular Location: base pairs 1,335,278 to 1,349,142 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- dishevelled-1
- dishevelled 1 (homologous to *Drosophila dsh*)
- dishevelled, *dsh* homolog 1
- DRS2
- DSH homolog 1
- DVL
- DVL1L1
- DVL1P1
- segment polarity protein dishevelled homolog DVL-1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The Wnt Gene Family and the Evolutionary Conservation of Wnt Expression
<https://www.ncbi.nlm.nih.gov/books/NBK6212/>

GeneReviews

- Autosomal Dominant Robinow Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK268648>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DVL1%5BTIAB%5D%29+OR+%28dishevelled+segment+polarity+protein+1%5BTIAB%5D%29%29+OR+%28dishevelled+1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- DISHEVELLED 1
<http://omim.org/entry/601365>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/DVL1ID463ch1p36.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DVL1%5Bgene%5D>
- HGNC Gene Family: Dishevelled segment polarity proteins
<http://www.genenames.org/cgi-bin/genefamilies/set/505>
- HGNC Gene Family: PDZ domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1220>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3084
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1855>
- UniProt
<http://www.uniprot.org/uniprot/O14640>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/DVL1>

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